Support for Senate Bill 465: An Act Requiring Newborn Screening For X-linked Adrenoleukodystrophy (ALD)

Senator Gerratana, Representative Johnson, and Public Health Committee Members

Thank you for the opportunity to speak in support of the Senate Bill 465. My name is Ann Moser. I am currently a research associate at the Kennedy Krieger Institute and the Dept. of Neurology at Johns Hopkins Medical School in Baltimore, Maryland. My late husband, Dr. Hugo Moser, and I developed an interest in studying ALD in 1978 when the group at Albert Einstein in NYC reported that patients with ALD had increased very long chain fatty acids (VLCFA) mainly of 26 carbons chain length (C26:0) in brain and adrenal cholesterol esters. In the early 1980's Hugo's research team at the Kennedy Krieger Institute developed gas chromatographic assays of the very long chain fatty acids, first in cultured cells and later in plasma, to diagnose patients with ALD. After the plasma C26:0 assay became available, many families with ALD were identified and therapy trials began. One of the most important, and available life-saving therapies for ALD is **hormone replacement** for those ALD patients with Addison's disease.

Since the early 1990's, **bone marrow transplantation** was shown to be effective in halting the central nervous system demyelination if done at the **first signs of progressive brain dysfunction.** By 2010 several hundred ALD boys have benefited from bone marrow and umbilical cord cell transplantation as well treatment for their Addison's disease.
It was Hugo Moser's dream to identify boys with ALD early, at a time before Addison's disease and brain dysfunction occurred. In 2005 Hugo suggested to the national newborn screening committee that ALD be added to the list of disorders that would possibly benefit from newborn screening, however, at that time there was no test for ALD utilizing the sample collected on all newborns, the heel stick blood spot on filter paper.

In order to develop a newborn test for ALD, Hugo and I contacted Walter Hubbard, Ph.D. at the Dept. of Clinical Pharmacology at Johns Hopkins. Walter is an expert in liquid chromatography tandem mass spectroscopy (LC/MSMS) of lipids and he was interested in helping us devise a test for ALD utilizing the newborn dried whole blood spot (DBS). In January of 2006, we determined that the C26 content of the lyso phosphatidylcholines (lyso PC) was 5 to 10 fold higher in whole venous blood spots from ALD patients when compared with controls. The ALD newborn DBS had a 5 to 15 fold increased C26:0 lyso PC with no overlap when compared with 500 anonymous newborn DBS. These findings were published in 2009. Since that time we have developed a high throughput LC/MSMS screening procedure and have published a combined extraction of the C26:0 lyso PC with that of the acyl carnitines. Recently together with the MD State Newborn Screening Lab, we have completed the screening of 5000 consented newborns born in 3 local Baltimore hospitals and did not find one positive, thus we believe that using our procedure the false positive rate will be low.

We are here today on the behalf of all ALD researchers, the ALD support groups who have donated funds to ALD newborn screening, and many ALD families worldwide to request that ALD be added to the uniform panel of screening tests performed on all newborns.

Thank you for your time and consideration of this important, life-saving request.

Ann B. Moser, Kennedy Krieger Institute, 707 North Broadway, Baltimore, MD 21205, tel: 443-923-2761, mosera@kennedykrieger.org